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Schizophrenia and Di George syndrome. Report of a case

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Case Report

Di George syndrome is the most frequent interstitial deletion of chromosome 22 in the q11 region. It has a prevalence of 1 in 6000 live births. Common physical phenotypic features include facial dysmorphology, cleft palate, congenital heart disease, hypoplastic or aplastic thymus and/or parathyroid gland. Nearly 40–80% of patients with Di George syndrome were found to have a psychiatric disorder, 25–30% fulfilling DSM-IV diagnostic criteria for schizophrenia. We report a case of a patient with Di George syndrome and schizophrenia.

Mr D. is a 22 year old single white man with Di George syndrome, diagnosed with genetic control (FISH method. Loss of thesis N25 in chromosome 22). From the age of 17 he had a history of psychiatric symptoms with poor outcome although he received several trials with different antipsychotic agents in monotherapy (atypical and typical). He had multiple verbal auditory and visual hallucinations, delusional ideas of grandiosity, reference and percecution, Capgras and Fregoli syndrome, motor restlessness, agitation, aggression, sleep disturbances and serious concentration difficulties. The total PANSS score was 146. He received a combination of two atypical agents (risperidone and quetiapine) in high doses and after twelve weeks the total PANSS score reduced to 99 (32% reduction).

Discussion

Nearly 25–30% of patients with Di George syndrome fulfill DSM-IV criteria for schizophrenia. These subgroup of patients seems to have some special characteristics such as a) late age of onset, b) IQ scores between 50–100 (with the vast majority between 70–80), c) poor social interaction, d) history of delayed speech and language development, e) high rates of schizotypy traits, f) less frequent negative symptoms and g) reduction in whole brain volumes, particularly white matter.

Conclusion

Di George syndrome seems to have a characteristic behavioral phenotype. Patients with Di George syndrome and schizophrenia may consist a different clinical subtype of schizophrenia.

References

1. Murphy KC: **The behavioural phenotype in velo-cardio-facial syndrome.** *Journal of Intellectual Disability Research* 2004, **48:**524-530.
2. Murphy KC, Jones LA, Owen MJ: **High rates of schizophrenia in adults with velo-cardio-facial syndrome.** *Archives of General Psychiatry* 1999, **56:**940-945.
3. Scambler PJ: **The 22q11 delation syndromes.** *Human Molecular Genetics* 2000, **9:**2421-2426.